

# Testing Options for Fabry Disease



Daisy and Viviana,  
Fabry patients

# WHY GET TESTED FOR FABRY DISEASE?

Fabry disease is inherited. If one family member is diagnosed with the disease, others are likely to be affected as well. If you know Fabry disease runs in your family, here are some reasons to consider getting tested:

- Reduce the diagnostic delay, because Fabry disease is progressive, meaning it can get worse over time
- Eliminate uncertainty
- Help make sense of previously unexplained symptoms
- The earlier Fabry disease is diagnosed, the earlier disease management can begin

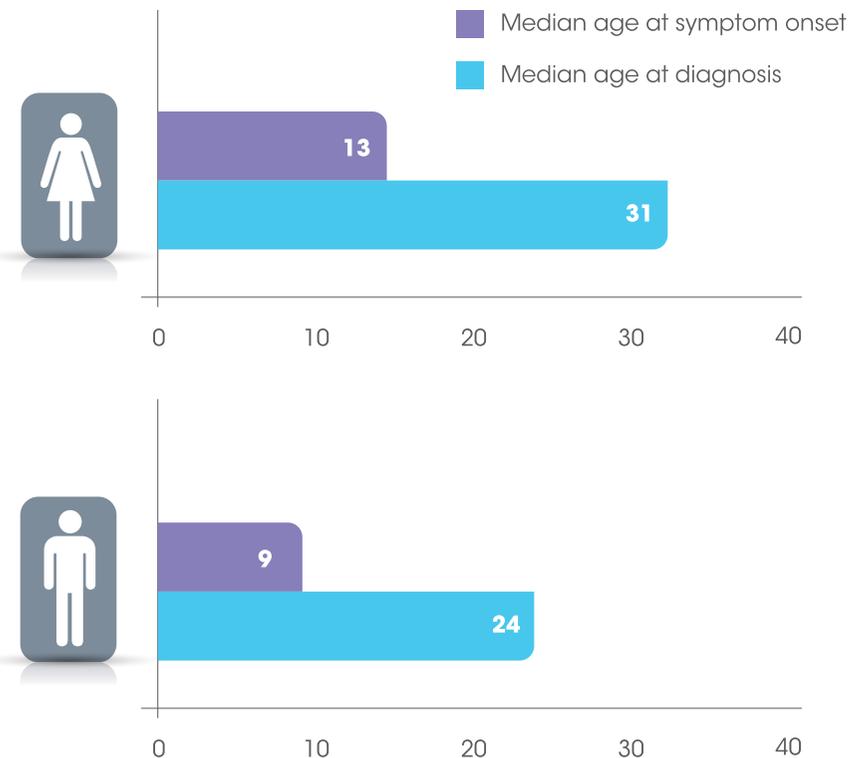
George, a Fabry patient



## Understanding the Diagnostic Journey

Prior to a diagnosis of Fabry disease, individuals may experience many years of suffering and frustration while potentially receiving unnecessary medical treatments due to misdiagnoses. Diagnosis of Fabry disease may be delayed by many years from when symptoms first appear. Many people see a number of different specialists before they get an accurate diagnosis, including:

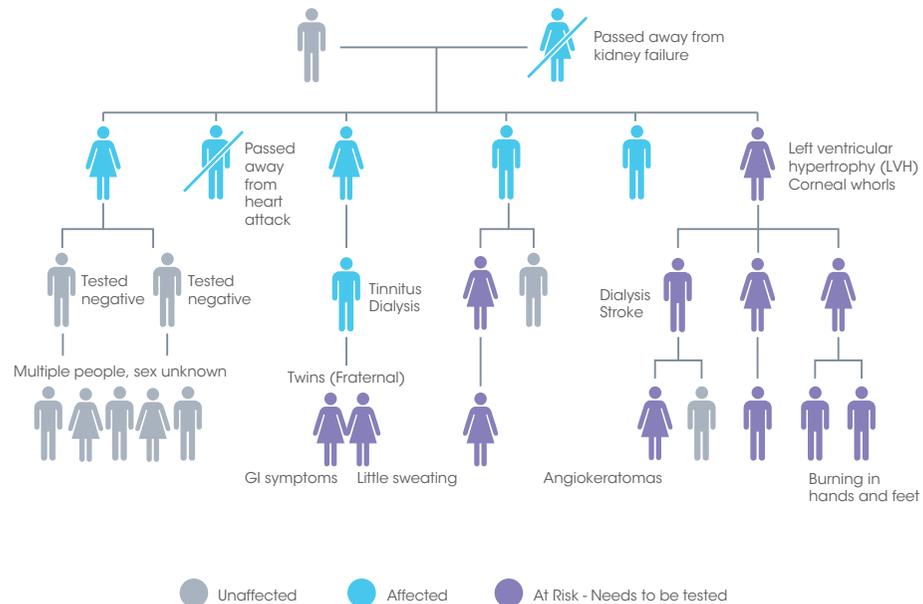
- Nephrologist for kidney problems
- Cardiologist for heart problems
- Neurologist for cerebrovascular problems, such as stroke
- Doctors for pain or gastrointestinal (GI) problems



## The First Step: Creating a Medical Family Tree

When one member of a family is diagnosed with Fabry disease, a medical family tree can help identify others who may be at risk.

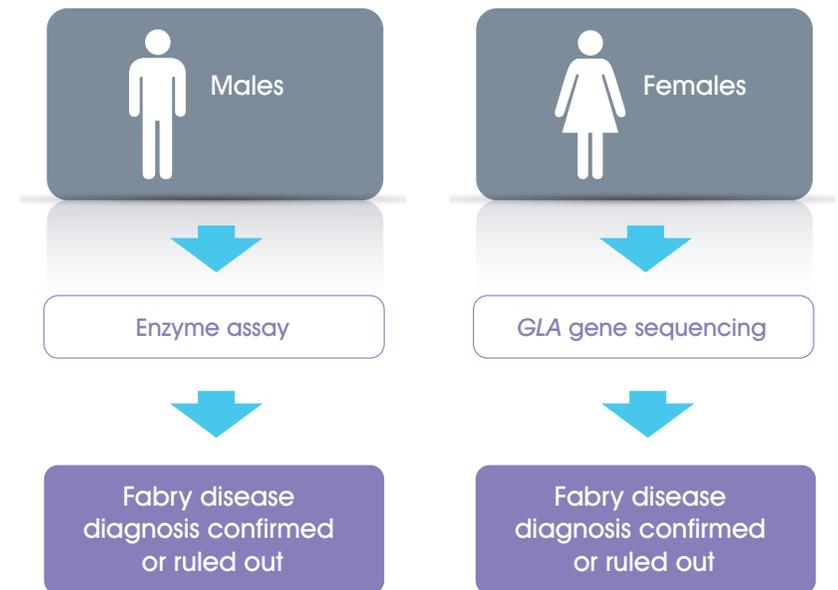
In this example, if the male on the lower right gets tested and learns that he has Fabry disease, it could help explain why his grandmother had left ventricular hypertrophy (enlarged left chamber of the heart). It is important to remember, though, that the absence of early signs and symptoms does not mean that a person is not affected. The disease can progress silently. Someone who inherits the altered gene is at risk for later complications, including heart disease, kidney disease and stroke.



A CareConnectPSS Patient Education Liaison can assist in creating a medical family tree.

## About Testing

- Fabry disease can be confirmed using a blood or saliva sample
- Many genetic labs around the country are able to analyze blood or saliva samples to diagnose Fabry disease
- You can simply have your blood or saliva sample sent to the lab; some doctors' offices are able to help with the blood draw, or you may go to a special blood draw center
- If you choose to be tested, you could have results in two months or less
- A genetic counselor can help before, during or after testing



- Males with Fabry disease usually have a very low amount of an enzyme called alpha-galactosidase
- A test called an enzyme assay is usually enough to confirm or rule out a diagnosis; it can be done with a blood sample
- Females may have either low or near normal enzyme levels, so they need to have gene sequencing, which can be done using a blood or saliva sample
- Gene sequencing can determine the family mutation, which is useful for screening other family members; diagnosed males may choose to have gene sequencing done for this reason

## Testing Options

Labs across the United States offer diagnostic tests for Fabry disease, and some provide the service free of charge. Below are some options available to you and your physician. This is not an all inclusive list. For additional testing centers that perform diagnostic testing, please visit <https://www.ncbi.nlm.nih.gov/gtr/>

Facility	Free Service*	Timing	Contact Information for Your Doctor
Duke University	No	Up to 2 weeks	919-613-8400 <a href="https://testcatalog.duke.edu">https://testcatalog.duke.edu</a>
Fabry Diagnostic Project (performed at EGL)	Yes	Up to 4 weeks	404-778-8570 <a href="http://www.genetics.emory.edu">www.genetics.emory.edu</a>
Greenwood Genetic Center	No	Up to 4 weeks	800-473-9411 <a href="http://www.ggc.org">www.ggc.org</a>
LabCorp/Integrated Genetics	No	Up to 3 weeks	<b>LabCorp Customers:</b> 800-345-4363 <a href="http://www.labcorp.com">www.labcorp.com</a> <b>Integrated Genetics Customers:</b> 800-848-4436 <a href="http://www.integratedgenetics.com">www.integratedgenetics.com</a>
The Lantern Project (performed at Perkin Elmer Genomics)	Yes	Up to 3 weeks	866-354-2910 <a href="http://www.LanternProjectDx.com">www.LanternProjectDx.com</a>
Mayo Medical Laboratories	No	Up to 3 weeks	800-533-1710 <a href="http://www.mayomedicallaboratories.com">www.mayomedicallaboratories.com</a>
Mount Sinai International Center for Fabry Disease	Yes	Up to 2 weeks	866-322-7963 <a href="http://icahn.mssm.edu/research/fabry">icahn.mssm.edu/research/fabry</a>
Sema4	No	Up to 3 weeks	800-298-6470 <a href="http://www.sema4.com">www.sema4.com</a>

\*In free testing programs, patients may be responsible for nominal costs such as the cost of the blood draw and overnight shipping to the lab.

This list of testing programs and/or laboratories is provided for informational use only. It should not be considered a recommendation or endorsement of any company, test or service for Fabry disease. Physicians are directed to contact the individual testing programs and/or laboratories for information on obtaining test requisitions, sample requirements, turnaround time and the specific details of the services provided. For commercial lab testing, appropriate coding and obtaining reimbursement is the responsibility of the provider submitting a claim for the item or service.

## Sanofi Genzyme Offers Support Services for You and Your Family

For more than 30 years, Sanofi Genzyme has been committed to helping meet the needs of people who are living with rare disorders like Fabry disease. Our commitment extends to those who have Fabry disease and their families. We offer a wide spectrum of services, all personalized and tailored to you through your Case Manager.

### Contact a CareConnectPSS Case Manager:

1-800-745-4447 (option 3)

Monday–Friday 8am–6pm EST

### Connect with us online:

[careconnectpss.com](http://careconnectpss.com)



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